

## **Evaluation of abnormal head size (Hydrocephalus ,microcephaly and craniocynostosis)**

### **objectives**

1. Recall the normal head growth from infancy to childhood.
2. Enumerate the common abnormalities in head size.
3. Define microcephaly.
4. Discuss and classify the different etiologies behind microcephaly.
5. Identify the key component from the history and physical examination to formulate the differential diagnosis, including history of pregnancy, neonatal and post natal period.
6. Explore the initial evaluation step of an infant with microcephaly.
7. Define macrocephaly.
8. Discuss and categorize the various causes of macrocephaly.
9. Describe the initial laboratory investigations and radiological imaging required while evaluating a child with macrocephaly.
10. Define and discuss the different types of hydrocephalus.
11. Describe and categorize the various etiologies of hydrocephalus.
12. Discuss the clinical presentation of hydrocephalus depending on the age.
13. Identify the component from the physical examination that aids in the diagnosis.
14. Outline the in initial evaluation steps and discuss management strategies of hydrocephalus.
15. Identify the indications for urgent CT scan imaging.
16. Outline the in initial evaluation steps and discuss management strategies of Craniocynostosis.

### **Head size**

- The measurement of head circumference (occipitofrontal circumference [OFC]), a direct reflection of head growth
- Important step in the evaluation of childhood growth and development.

- Accurate head circumference (HC) measurement is obtained with a flexible non-stretchable measuring tape pulled tightly across the most prominent part on the back (occiput) and front (supraorbital ridges) of the head.

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- First three months of life → the infants will gain six centimeters
  - 2 centimeters per month.
- Next three months → three centimeters in three months
  - 1 cm per month.
- Next six months → three centimeters in six months
  - 0.5 cm per month.
- Deviations from normal head growth may be the first indication of an underlying congenital, genetic, or acquired problem.

### **Microcephaly**

- A head circumference that measures more than 3 standard deviations below the mean for age and sex.

### **Etiologies of congenital microcephaly**

- Genetic
  - Isolated
    - Autosomal recessive microcephaly
    - Autosomal dominant microcephaly
    - X-linked microcephaly
    - Chromosomal (rare: “apparently” balanced rearrangements and ring chromosomes)
- Genetic

|  |
|--|
| <b><i>Syndromic</i></b>                    |
| <b><i>Chromosomal</i></b>                  |
| Trisomy 21, 13, 18                         |
| Unbalanced rearrangements                  |
| <b><i>Contiguous gene deletion</i></b>     |
| 4p deletion (Wolf-Hirschhorn syndrome)     |
| 5p deletion (cri-du-chat syndrome)         |
| 7q11.23 deletion (Williams syndrome)       |
| 22q11 deletion (velocardiofacial syndrome) |
| <b><i>Single gene defects</i></b>          |
| Cornelia de Lange syndrome                 |
| Holoprosencephaly (isolated or syndromic)  |
| Smith-Lemli-Opitz syndrome                 |
| Seckel syndrome                            |

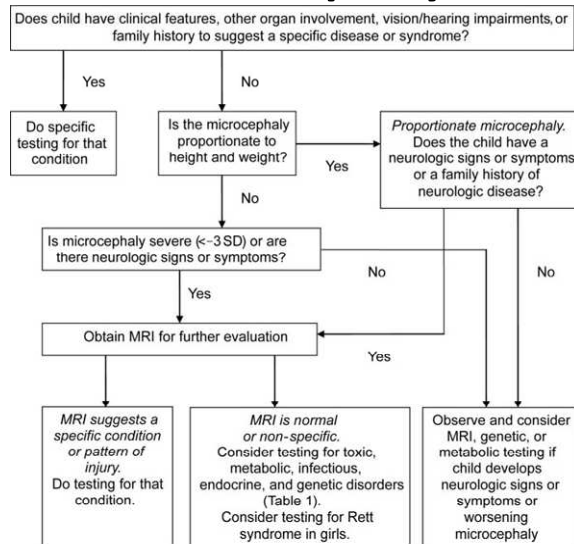
Etiologies of post natal onset microcephaly

|   |
|---|
| <i>Contiguous gene deletion</i>           |
| 17p13.3 deletion (Miller-Dieker syndrome) |
| <i>Single gene defects</i>                |
| Rett syndrome                             |
| Nijmegen breakage syndrome                |
| Ataxia-telangiectasia                     |
| Cockayne syndrome                         |
| Aicardi-Goutieres syndrome                |
| XLAG syndrome                             |
| Cohen syndrome                            |

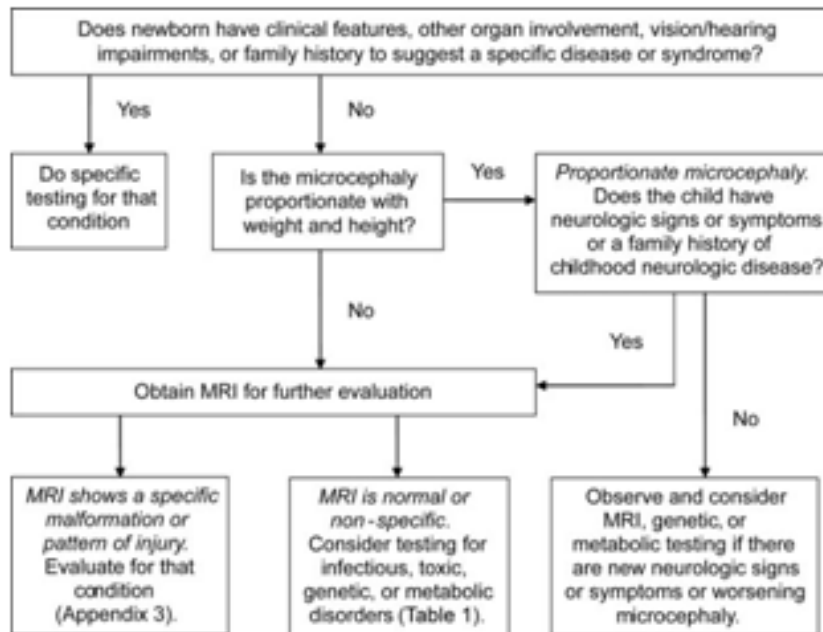
acquired post natal microcephaly

|                                 |
|---------------------------------|
| <i>Disruptive injuries</i>      |
| Traumatic brain injury          |
| Hypoxic-ischemic encephalopathy |
| Hemorrhagic and ischemic stroke |
| <i>Infections</i>               |
| Meningitis and encephalitis     |
| Congenital HIV encephalopathy   |
| <i>Toxins</i>                   |
| Lead poisoning                  |
| Chronic renal failure           |
| <i>Deprivation</i>              |
| Hypothyroidism                  |
| Anemia                          |
| Malnutrition                    |
| Congenital heart disease        |

# Evaluation of postnatal onset microcephaly



# Evaluation of congenital microcephaly



## Macro-cephaly

- OFC greater than two standard deviations (SD) above the mean for a given age, sex, and gestation (i.e.,  $\geq 97$ th percentile)
- Megalencephaly (also called macrencephaly) is enlargement of the brain parenchyma

## etiology

- Increased brain (megalencephaly)
  - Anatomic
    - Familial megalencephaly
    - Neurocutaneous disorders (eg, neurofibromatosis)
    - Autism spectrum disorder

- Achondroplasia
- Cerebral gigantism (Sotos syndrome)
- Fragile X syndrome
- PTEN hamartoma syndromes (eg, Cowden syndrome, Bannayan-Riley-Ruvalcaba)
- Increased brain (megalocephaly)
- Metabolic
  - Leukodystrophies (eg, Alexander, Canavan, megalencephalic leukoencephalopathy)
  - Lysosomal storage disorders (eg, Tay-Sachs, mucopolysaccharidosis, gangliosidosis)
- Increased cerebrospinal fluid
  - Hydrocephalus
  - Benign enlargement of the subarachnoid space
  - Hydranencephaly
  - Choroid plexus papilloma
- Increased blood
  - Hemorrhage
  - Intraventricular
  - Subdural
  - Epidural
  - Subarachnoid
  - Arteriovenous malformation
- Increased bone
  - Bone marrow expansion
    - Thalassemia major
  - Primary bone disorders

- Skeletal and cranial dysplasias
  - Achondroplasia
  - Osteogenesis imperfecta
  - Cleidocranial dysostosis
  - Metaphyseal dysplasia
  - Osteopetrosis
  - Hyperphosphatasia
- Increased intracranial pressure
  - Idiopathic (pseudotumor cerebri)
  - Infection or inflammation (eg, meningitis)
  - Toxins
    - Lead
  - Metabolic abnormalities
    - Vitamin A deficiency or excess
    - Galactosemia
- Mass lesions
  - Intracranial cyst.
  - Intracranial tumor.
  - Intracranial abscess.

### **Hydrocephalus**

- Not a specific disease
- Represents a diverse group of conditions that result from
  - Impaired circulation and absorption of CSF
  - Increased production of CSF



## CSF production

- Formed primarily in the ventricular system by the choroid plexus
  - Situated in the lateral, 3rd, and 4<sup>th</sup> ventricles.
  - Most CSF is produced in the lateral ventricles.
- ≈ 25% originates from extrachoroidal sources
  - capillary endothelium within the brain parenchyma.
- CSF production rate in a normal child is about 20 mL/hr of
- The total volume of CSF approximates
  - 50 mL in an infant
  - 150 mL in an adult.
- Most of the CSF is extraventricular.

## CAUSES OF HYDROCEPHALUS

- COMMUNICATING
  - Achondroplasia
  - Basilar impression
  - Benign enlargement of subarachnoid space
  - Choroid plexus papilloma
  - Meningeal malignancy
  - Meningitis
  - Posthemorrhagic
- NONCOMMUNICATING
  - Aqueductal stenosis
  - Infectious\*
  - X-linked
  - Mitochondrial

- Autosomal recessive
- Autosomal dominant
- L1CAM mutations
- Chiari malformation
- Dandy-Walker malformation
- Klippel-Feil syndrome
- Mass lesions
- Abscess
- Hematoma
- Tumors and neurocutaneous disorders
- Vein of Galen malformation
- Walker-Warburg syndrome
- HYDRANENCEPHALY
  - Holoprosencephaly
  - Massive hydrocephalus
  - Porencephaly

#### CLINICAL MANIFESTATIONS

- Variable
- Depends on many factors, including
  - The age at onset.
  - The nature of the lesion causing obstruction.
  - The duration and rate of increase of the intracranial pressure (ICP).

#### In an infant

- An accelerated rate of enlargement of the head
- Anterior fontanel

- Wide open and bulging
- Dilated scalp veins.
- Broad forehead
- Setting-sun eye sign
  - The eyes might deviate downward because of impingement of the dilated suprapineal recess on the tectum.
- Long-tract signs
  - Owing to stretching and disruption of the corticospinal fibers.

In an older child

- The cranial sutures are partially closed
  - The signs may be subtler.
- Irritability, lethargy, poor appetite, and vomiting
  - Common to both age groups.
- Headache
  - Prominent symptom in older patients.
- A gradual change in personality and deterioration in academic productivity suggest a slowly progressive form of hydrocephalus.
- Serial measurements of the head circumference often indicate an increased velocity of growth.
- Percussion of the skull might produce a cracked pot sound or MacEwen's sign
  - Separation of the sutures.
- A foreshortened occiput suggests Chiari malformation
- Prominent occiput suggests the Dandy-Walker malformation.
- Papilledema, abducens nerve palsies, and pyramidal tract signs, which are most evident in the lower extremities, are apparent in many cases.

Diagnosis: history

- Familial cases
  - X-linked or autosomal hydrocephalus secondary to aqueductal stenosis.

- A past history
  - Prematurity with intracranial hemorrhage
  - Meningitis, or mumps encephalitis
- Multiple café-au-lait spots and other clinical features of neurofibromatosis point

Aqueductal stenosis as the cause of hydrocephalus

Examination

Careful inspection, palpation, and auscultation of the skull and spine.

- The occipitofrontal head circumference is recorded and compared with previous measurements.
- The size and configuration of the anterior fontanel
- The back is inspected for abnormal midline skin lesions, including tufts of hair, lipoma, or angioma
  - Spinal dysraphism.
- The presence of a prominent forehead or abnormalities in the shape of the occiput can suggest the pathogenesis of the hydrocephalus.
- A cranial bruit is audible in association with many cases of vein of Galen arteriovenous malformation.
- Transillumination of the skull is positive with massive dilatation of the ventricular system or in the Dandy-Walker syndrome.
- Inspection of the eye grounds
  - Chorioretinitis suggests an intrauterine infection, such as toxoplasmosis
  - Papilledema is observed in older children
  - Rarely present in infants

Work up

- Plain skull films typically show
  - Separation of the sutures
  - Erosion of the posterior clinoids in an older child, and an increase in convolutional markings
    - (beaten-silver appearance) with longstanding increased ICP.

- The CT scan and/or MRI along with ultrasonography in an infant are the most important studies to identify the specific cause and severity of hydrocephalus.

## DIFFERENTIAL DIAGNOSIS

- Megalencephaly
- Hydranencephaly
  - The cerebral hemispheres are absent or represented by membranous sacs with remnants of frontal, temporal, or occipital cortex dispersed over the membrane.
  - The midbrain and brainstem are relatively intact
  - Cause: unknown
    - Bilateral occlusion of the internal carotid arteries during early fetal development

## TREATMENT

- Depends on the cause
- Medical management (acetazolamide and furosemide)
  - Temporary relief by reducing the rate of CSF production
  - Long-term results have been disappointing
- Most cases require extracranial shunts
  - Ventriculoperitoneal shunt.
  - Endoscopic third ventriculostomy (ETV)

## PROGNOSIS

- Depends on the cause
- Increased risk for
  - Various developmental disabilities
  - Abnormalities in memory function
  - Ophthalmology problems including
    - Strabismus

- Visuospatial abnormalities
- Visual field defects
- Optic atrophy with decreased acuity
- Accelerated pubertal development in patients with shunted hydrocephalus or myelomeningocele
- It is imperative that hydrocephalic children receive long-term follow-up in a multidisciplinary setting.

### **Craniosynostosis**

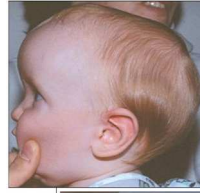
- Premature closure of the cranial sutures
- Classified as
  - Primary
    - Closure of one or more sutures owing to abnormalities
    - Incidence approximates 1/2,000 births
  - Secondary
    - Results from failure of brain growth and expansion
- Associated with varying types of abnormal skull shape.
- The cause is unknown in the majority of children
  - Genetic syndromes account for 10-20%

### **CLINICAL MANIFESTATIONS**

- Most cases are evident at birth
- Characterized by a skull deformity that is a direct result of premature suture fusion.
- Palpation of the suture reveals a prominent bony ridge

## Scaphocephaly

- Long and narrow skull.
- Premature closure of the sagittal suture.
- **The most common form of craniosynostosis.**
- Sporadic.
- More common in males.
- Often causes difficulties during labor.
- Does not produce increased ICP or hydrocephalus.
- Neurologic examination of affected patients are normal.



## Frontal plagiocephaly

- **Next most common form of craniosynostosis.**
- Unilateral flattening of the forehead, elevation of the ipsilateral orbit and eyebrow, and a prominent ear on the corresponding side.
- More common in females .
- Result of premature fusion of a coronal and sphenofrontal suture.
- Surgical intervention
  - Cosmetically pleasing result.
- When imaging does not reveal a closed suture
  - Positional factors are of primary importance.



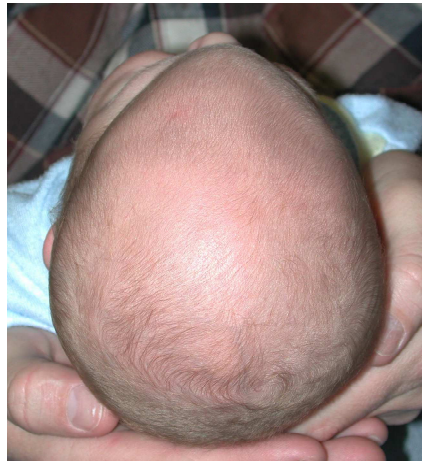
## Occipital plagiocephaly

- **Most often a result of positioning** during infancy.
- More common in an immobile child or a child with a disability.
- Fusion or sclerosis of the lambdoid suture can cause unilateral occipital flattening and bulging of the ipsilateral frontal bone.



## Trigonocephaly

- **Rare form of** craniosynostosis
- Premature fusion of the metopic suture.
- These children have a keel-shaped forehead and hypotelorism
- Risk for associated developmental abnormalities of the forebrain.
- Milder forms of metopic ridging are more common.





# Turriccephaly

- **A cone-shaped head**
- To premature fusion of the coronal and often sphenofrontal and frontoethmoidal sutures.
- The **kleeblattschädel deformity** is a peculiarly shaped skull that resembles a cloverleaf.
- Very prominent temporal bones, and the remainder of the cranium is constricted.
- **Hydrocephalus is a common**



- Premature fusion of only one suture rarely causes a neurologic deficit.
  - The sole indication for surgery is to enhance the child's cosmetic appearance.
  - The prognosis depends on the suture involved and on the degree of disfigurement.
- Neurologic complications, including hydrocephalus and increased ICP, are more likely to occur when two or more sutures are prematurely fused, in which case operative intervention is essential.

## syndromic

- Each of the genetic syndromes poses a risk of additional anomalies, including
  - Hydrocephalus
  - Increased ICP
  - Papilledema
  - Optic atrophy
  - Respiratory problems secondary
    - Deviated nasal septum

- Choanal atresia
  - Disorders of speech and deafness.
- Craniectomy is mandatory for management of increased ICP
- A multidisciplinary craniofacial team is essential for the long-term follow-up of affected children.  
Craniosynostosis
- Surgically corrected
  - Good outcomes
  - Relatively low morbidity and mortality, especially for nonsyndromic infants.